



TGIF1 gene

TGFB induced factor homeobox 1

Normal Function

The *TGIF1* gene provides instructions for making a protein called TG-interacting factor. This protein is important for normal development of the front part of the brain (forebrain). TG-interacting factor is a transcription factor, which means that it regulates the activity of certain genes. This protein turns off genes by attaching (binding) to specific regions of DNA or by interacting with other DNA-binding proteins.

TG-interacting factor regulates signaling pathways that are important for embryonic development. This protein blocks the signals of the transforming growth factor beta (TGF- β) pathway. This signaling pathway transmits chemical signals from the cell surface to the nucleus, which allows the environment outside the cell to affect how the cell produces other proteins. TG-interacting factor also blocks a molecule called retinoic acid from regulating gene activity. Retinoic acid, a form of vitamin A, binds to a group of transcription factors that regulate a number of genes important for early development. By blocking these signaling pathways, TG-interacting factor ensures that certain genes are turned off at the proper time.

Health Conditions Related to Genetic Changes

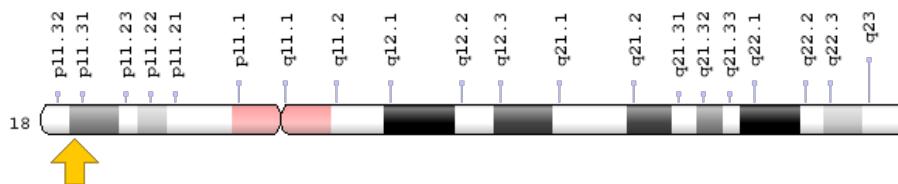
nonsyndromic holoprosencephaly

At least 13 mutations in the *TGIF1* gene have been found to cause nonsyndromic holoprosencephaly. This condition occurs when the brain fails to divide into two halves (hemispheres) during early development. *TGIF1* gene mutations are the fourth most common cause of nonsyndromic holoprosencephaly. These mutations disrupt the protein's ability to bind with DNA or interact with other proteins. As a result, TG-interacting factor cannot block the signals of the TGF- β pathway and retinoic acid. If the signals involved in forebrain development are not properly regulated, the brain does not separate into two hemispheres. The signs and symptoms of nonsyndromic holoprosencephaly are caused by abnormal development of the brain and face.

Chromosomal Location

Cytogenetic Location: 18p11.31, which is the short (p) arm of chromosome 18 at position 11.31

Molecular Location: base pairs 3,411,927 to 3,458,412 on chromosome 18 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- 5'-TG-3' interacting factor
- 5'-TG-3'-interacting factor 1
- homeobox protein TGIF
- homeobox protein TGIF1
- HPE4
- MGC5066
- MGC39747
- TALE homeobox TG-interacting factor
- TGFB-induced factor homeobox 1
- TGIF
- TGIF1_HUMAN
- transforming growth factor-beta-induced factor

Additional Information & Resources

Educational Resources

- Neuroscience (second edition, 2001): The Molecular Basis of Neural Induction
<https://www.ncbi.nlm.nih.gov/books/NBK10823/>

GeneReviews

- Holoprosencephaly Overview
<https://www.ncbi.nlm.nih.gov/books/NBK1530>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28TGIF1%5BTIAB%5D%29+OR+%28%28HPE4%5BTIAB%5D%29+OR+%28TGIF%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D>

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<http://omim.org/entry/602630>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_TGIF1.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=TGIF1%5Bgene%5D>
- HGNC Gene Family: TALE class homeoboxes and pseudogenes
<http://www.genenames.org/cgi-bin/genefamilies/set/526>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=11776
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/7050>
- UniProt
<http://www.uniprot.org/uniprot/Q15583>

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